# Surgical Treatment of Scoliosis in a Spinal Muscular Atrophy Population

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**Seventy-eight patients were diagnosed with spinal muscular atrophy between 1969 and 1988. Scoliosis developed in 34 of these patients, an incidence of 60%. Thirty-one patients could be retrospectively reviewed by chart review or interview. The average follow-up was 11.5 years. Onset of scoliosis averaged 8.8 years. Twenty-two patients were treated nonsurgically and nine surgically. Patients had improved sitting balance and endurance after surgery. Complications of surgery included loss of correction in one patient, one pseudarthrosis, and one patient who required prolonged ventilatory support. The prolonged survival of patients with spinal muscular atrophy justifies aggressive orthopaedic management of scoliosis to prevent progression of deformity and improve sitting comfort. [Key words: spinal muscular atrophy, paralytic scoliosis]**

S PINAL MUSCULAR ATROPHY is an autosomal-recessive neuromuscular disorder of childhood. It was first described by Werdnig and Hoffman in the early 1890s. The Werdnig-Hoffman form of spinal muscular atrophy (SMA) is also known as infantile SMA. It presents in the first few months of life, and death occurs within the first 2 years. A milder form was described by Kugelberg and Welander. In this juvenile form, the diagnosis is not made until the child is 2 years old and the prognosis for survival is improved. There is also an intermediate form that overlaps the juvenile form. A benign or arrested classic Werdnig-Hoffman has been described[7] in which the diagnosis is made in infancy but the clinical course is unexpectedly benign. It is believed that the different forms are in fact a continuum of the same disorder.[4] As a result, many classification systems, based on the severity and the prognosis, have been used to compare the children.[2,4,8,13] In general, it is agreed that the earlier the onset, the worse the prognosis.[4]

The pathology of SMA is degeneration of anterior horn cells of the spinal cord, which causes extensive weakness. The proximal muscles are more affected than the distal musculature, and the lower extremities are more affected than the upper extremities. Mild cases may have no arm involvement.[2] The trunk may also demonstrate weakness, but the degree of weakness does not necessarily correlate with the severity.[2] The neck and facial muscles may be weak, and the tongue has fasciculations. There is loss of deep tendon reflexes, but sensation is unaffected. Clinically, the disorder can be confused with other neuromuscular conditions such as Duchenne muscular dystrophy, limb-girdle muscular dystrophy, or poliomyelitis. The diagnosis is made on the clinical examination, electromyography, nerve conduction studies, and muscle and nerve biopsies.

The loss of anterior horn cells is considered an acute event without progression. The neurologic deterioration may stabilize and remain unchanged for long periods of time.[10] The progressive motor weakness that is demonstrated clinically is a reflection of normal growth that outpaces the marginal muscle reserve.[5] Respiratory function is compromised and atelectasis and pneumonia are the usual causes of death. Scoliosis has been reported to occur in all but the mildest cases[2] and is progressive once it develops.

A comprehensive review of all patients treated at our institution with the diagnosis of infantile SMA was undertaken with particular attention to the development of spinal deformity. Decisions about the most appropriate treatment of an established spinal curvature are difficult. The children frequently have respiratory compromise, and life expectancy is unpredictable.

## MATERIALS AND METHODS

There were 78 patients diagnosed with spinal muscular atrophy between 1969 and 1988. All the patients were younger than 12 years of age at the time of presentation, with an average age of 9.3 months. The diagnosis was made in all patients with neurologic examination that demonstrated the typical weakness, tremors, areflexia, and normal sensation. The diagnosis was confirmed with electromyography and muscle biopsy. Occasionally children will present with weakness that is presumed to be SMA but cannot be distinguished from other neuromuscular disorders such as limb-girdle or Duchenne muscular dystrophies. Children with uncertain etiologies of weakness were eliminated from the study. After the initial diagnosis, 57 patients continued to be managed at our institution. Follow-up consisted of clinical examinations, chart review, mail questionnaires, and/or telephone interviews. The average length of follow-up for these patients was 8.8 years.

Classification of the patients was attempted using the traditional Werdnig-Hoffman, Kugelberg-Welander, and intermediate forms of the disease. The chart review revealed that many children had been labeled with a particular form that did not correspond to the clinical course. To be consistent, patients were classified based on the age of diagnosis. Patients in group I presented at less than 6 months of age. Patients in group II were diagnosed between 6 months and 18 months of age. The remaining patients, group III, were not diagnosed until after age 18 months.

## RESULTS

Nineteen of the 57 patients died of respiratory and cardiac complications associated with SMA. The average time of death was 2.4 years (range, 3 months to 10 years). Eighteen of these patients were in the group with an early diagnosis and were severely involved (group I); the remaining death occurred in a group II patient. Four of these patients had spinal deformity documented before death and are included in the subgroup of patients that constitutes the focus for review. Double major curves (Figure 1A), thoracic curves, and sagittal plane deformities (Figures 2B and 1B). Severe rotation with chest wall deformity and razor back rib prominence frequently accompanied thoracic scoliosis. Twenty-two of the 31 patients were treated nonsurgically for their spine deformities, and nine patients underwent surgical treatment.
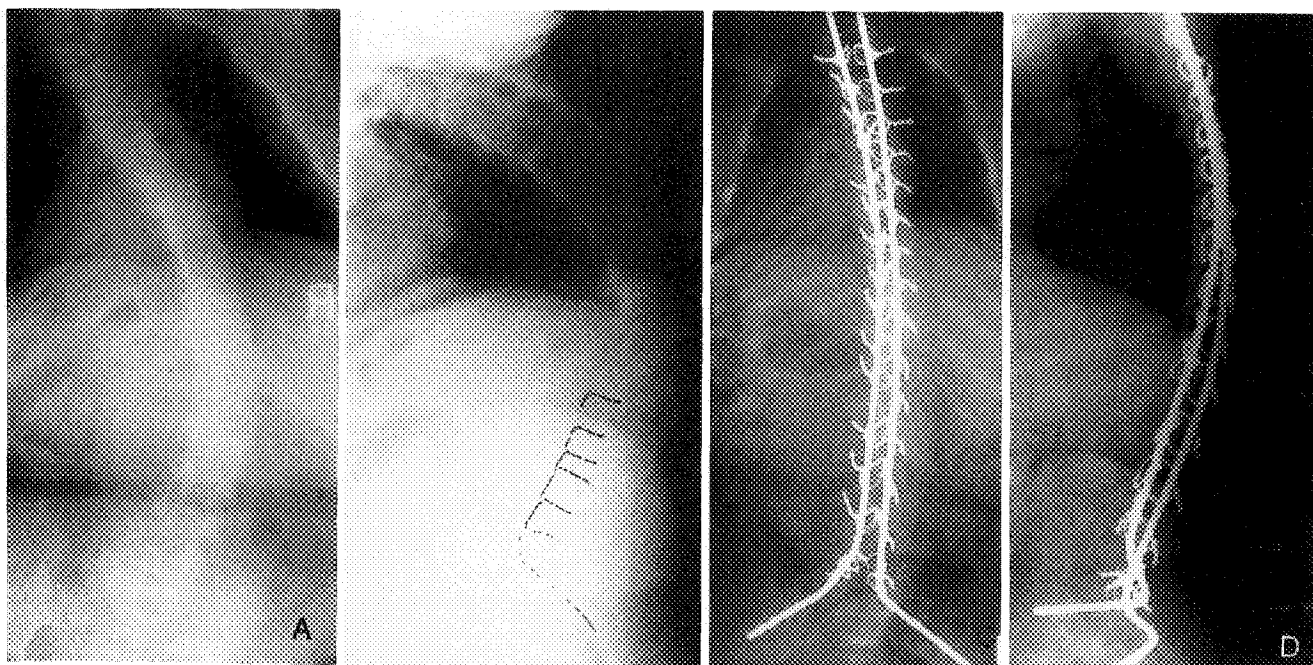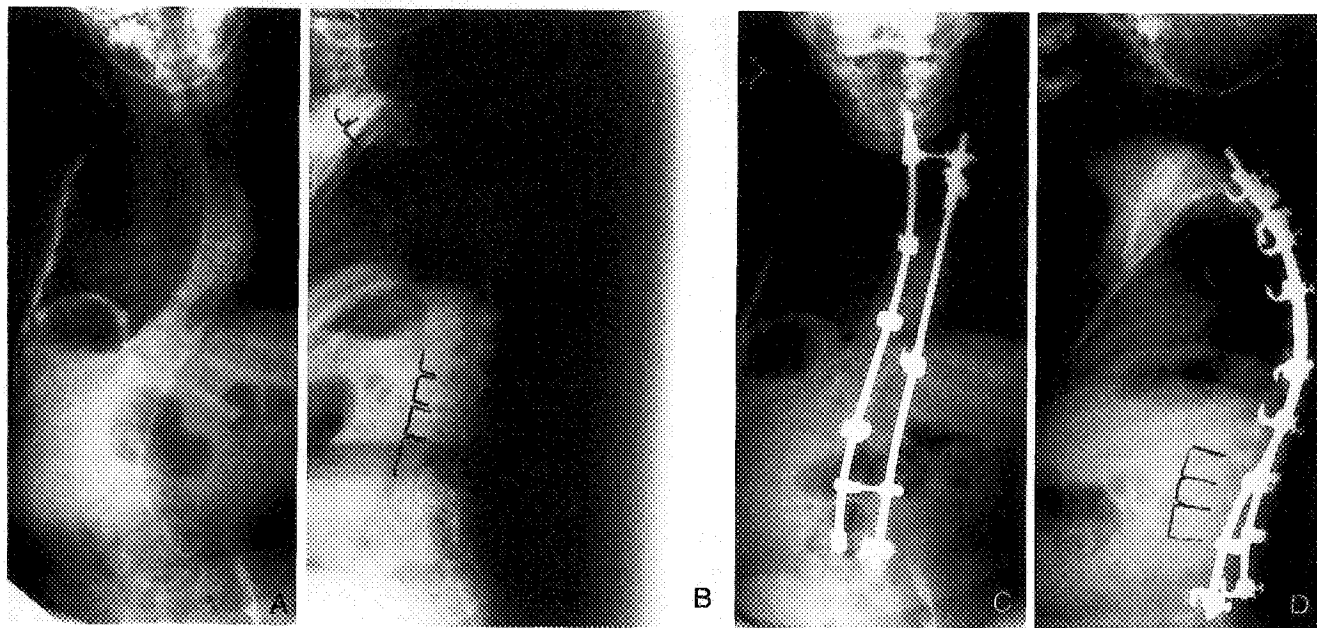
**Fig 1.** (A) Preoperative AP and (B) lateral radiographs of a double major curve pattern and upper thoracic kyphosis. (C) AP and (D) lateral postoperative radiographs. The patient underwent Cotrel-Dubousset instrumentation without fusion due to her young age. The curve has continued to progress despite instrumentation.

Of the 22 patients treated nonsurgically, 13 were classified as group I, 3 as group II, and 6 as group III. The age at onset of spinal deformity averaged 7.9 years for these patients (range, 4 to 13 years). The average length of follow-up was 10.7 years (range, 2.5 to 28 years). The age at final follow-up was 13.0 years (range, 2 to 42 years), or 14.8 (range, 4 to 42) if the patients that died are excluded.

Twenty-one of the patients who were members of the group treated conservatively had scoliosis, and one patient had a postural kyphosis. The scoliosis was mild (less than 20°) in 13 patients, and severe in 8.

There were eight patients who were treated in a brace. Six of the patients who were treated in braces had severe curves. A thoracolumbosacral orthosis (TLSO) was prescribed for two patients, and two were given Milwaukee braces to control their curves. One patient in a TLSO for a double major curve of 130°, and one patient in a Milwaukee brace for a 100° curve were unable to comply with brace treatment due to discomfort. There were two patients with minimal curves who were treated in braces because of their young ages (both 4 years of age); one of these patients died. Six of the patients with mild curves who were being



**Fig 2.** (A) Preoperative AP and (B) lateral radiographs in a patient with a long, C-shaped paralytic curve and significant kyphosis. (C) AP and (D) lateral radiographs of the same patient after Luque segmental instrumentation.

observed were younger than 14 years of age. Only longer follow-up will reveal the natural history of these curves. The remaining five patients with mild curves were older than 14 years of age at final follow-up.

Twelve of the patients who were treated nonsurgically had never been ambulatory. The remaining 10 patients were ambulatory in early childhood. Two patients had lost the ability to ambulate, one at age 8 years and one after a femoral fracture at age 12. Eight patients remained at least household ambulators at final follow-up.

The four deaths in the spinal deformity group occurred in the group treated nonsurgically. One patient died at the age of 4 years after 36 months of follow-up. She had documented scoliosis that did not require treatment. The second patient died at the age of 2 years after 19 months of follow-up with a documented right thoracic scoliosis. The third patient died at age 10, after 9.9 years of follow-up and brace treatment for a severe curve. The final death occurred in the 4-year-old who had been followed since birth and treated in a brace for a mild curve.

There were no complications related to brace wear. Two patients discontinued their braces due to discomfort but no significant skin complications were reported.

Nine patients underwent surgical correction for scoliosis. There were a total of 10 surgical procedures performed. Five patients had a diagnosis at less than 6 months and were classified as group I, two as group II (diagnosis at 6 to 18 months), and two as group III (diagnosis older than 18 months). The spinal deformity was first diagnosed at an average age of 9.5 years (range, 4 to 13 years). Before performing the surgery, the patients were followed for an average of 2 years (range, 1 to 4 years) after the diagnosis of scoliosis had been made. The patients were followed for an average of 5 years (range, 6 months to 13 years) after surgery. The average length of follow-up from the initial diagnosis of spinal muscular atrophy was 14.6 years (range, 5 to 23 years). The average age of the patients was 15.2 years at the time of final follow-up (range, 6 to 25 years).

Surgery was performed for curves that demonstrated progression, or for severe scoliosis. Five patients had preoperative curve measurements of 35 to 65°. Three patients had curve measurements of greater than 6° but less than 100°. One patient had a curve over 100° before surgical correction. Only one patient had previously been treated in a brace, and underwent surgery for a 65° curve. Instrumentation included four Harrington rods, five Luque rods (Figure 1), one subcutaneous Cotrel-Dubousset without fusion (Figure 2), and one Dwyer. One patient who had had a Harrington instrumentation and fusion underwent a revision using a Dwyer and a Luque for a pseudoarthrosis. No patient had preoperative traction.

None of the patients treated surgically were ambulatory at the time of surgery. Two patients had been ambulatory in childhood, but had become wheelchair dependent before surgery. Patients reported better sitting balance and improved comfort after surgery. Endurance was improved as a result. Sitting height was improved.

The complications included loss of correction in the child who had a subcutaneous rod without fusion. Surgery was performed at age 6 years for a 35° double major curve. At follow-up 6 months later, the curves had progressed to 42°. No other patient demonstrated significant loss of correction. One patient had a pseudarthrosis and required revision with an anterior fusion and Dwyer instrumentation and repeat posterior fusion. One patient had a painful prominence of the Luque rod, which was subsequently shortened, with relief of her symptoms. There was only one patient with significant postoperative respiratory compromise that required tracheostomy for prolonged ventilatory support. No patient had a wound complication. There were no deaths in the surgically treated group.

## DISCUSSION

Spinal muscular atrophy presents in infancy or early childhood. The severity of the disorder correlates with the age at diagnosis.[2] In children diagnosed in infancy, the prognosis for survival is poor. However, Schwentker[12] found that if the children survive beyond infancy they have a fair prognosis for survival into adulthood. Dubowitz[7] identified the arrested form of infantile SMA, in which children with early

diagnosis have a benign clinical course. Eighteen of the 31 patients in this study were diagnosed before age 6 months and were presumed to have a poor prognosis for survival. Severe curves developed in all but one of the patients in our series who were diagnosed with SMA at 6 months of age or younger. Five of the children treated surgically at an average age of 12.5 years had been diagnosed before the age of 6 months and given a poor prognosis for survival.

Prolonged survival has resulted in a high incidence of spine deformity. We had a 60% incidence of scoliosis in our population. Other studies have found a similar incidence of scoliosis and a higher incidence in the more severe forms of spinal muscular atrophy. Benady found a 54% incidence of scoliosis, with scoliosis complicating all the more severe forms of the disease.[2] Evans diagnosed scoliosis in all the severe cases of SMA, and in one half of the less severe cases. Schwentker reported a 70% incidence of scoliosis.

Children should be closely monitored for the development of scoliosis. The average age of diagnosis of scoliosis was close to 9 years in our group of patients, with the youngest 5 months of age and the oldest 14 years of age. Brown et al[3] noted that scoliosis developed in all of their patients before puberty; the youngest was diagnosed at 3 years of age. Evans found that the more severe the muscle weakness, the earlier the onset of scoliosis and the more progressive the spinal deformity.[8] Daher et al[5] reported a group of surgically treated patients who were diagnosed with spine deformity as early as at the time of birth to as old as 14 years, with an average age near 7 years.

The early onset and rapid progression of scoliosis in SMA is well recognized. There is still controversy about treatment modalities, indications for treatment, and the appropriate time to begin treatment. The potential benefits of bracing and surgery must be weighed against the complications of both treatments and the possibility of functional loss.

Eight of the patients in our study were treated with a brace. There were no complications related to the brace wear. Two patients who had flexible but severe curves were treated with braces but they discontinued the braces because of discomfort. Furumasu et al[9] found that patients treated in braces were less functional because of decreased flexibility of the spine and resulted in noncompliance. On the other hand, Benady[2] recommended early detection of scoliosis and bracing prophylactically to prevent scoliosis in the more severe forms of SMA. Attempts to brace before spinal deformity developed did not prevent scoliosis. Evans et al did not brace prophylactically but did use bracing early to slow progression of the curves and to aid in sitting balance. Shapiro[13] recommends bracing when the curve reaches 15 to 20°. Severe chest wall deformities have resulted from bracing[1,11] and may be a contraindication to continued brace treatment.

Bracing may not eliminate the need for surgical correction and stabilization of the deformity, but it may delay surgery until closer to the end of growth. A recent study has shown that there is a high incidence of curve progression, despite a posterior spinal fusion, when surgery performed in young children.[6] Surgery at a young age would require anterior and posterior approaches to prevent this "crankshaft phenomenon" with further growth. The advantage of bracing is that it may control the curve until a posterior fusion alone can be performed without the risk of continued progression.

Nine of the patients in this study underwent posterior spine fusion for progressive or severe curves. Only one of those patients had been treated preoperatively in a brace. Evans et al[8] recommend surgery when the curve reaches 60°. They performed spine fusion in children younger than 10 years of age if their curves had progressed to the 60° range to prevent severe deformity and compromised respiratory function. Shapiro and Bresnan[13] performed posterior spine fusion in patients as young as 7 years old if the curve reached 40 to 60°. Surgery was performed in our series when the curves were over 35°. Four patients

had curves that measured more than 65°. We believe that early stabilization prevents the severe chest wall deformity that can result from scoliosis and allows surgery to be performed before severe respiratory compromise increases the risk of surgery.

There is some evidence that spine fusions can decrease function.[1,3,8,9,12] Schwentker reported a loss of muscle strength and motor skills after spine surgery, although the comfort and stability of the back outweighed these disadvantages. It is uncertain what role prolonged recumbency after surgery played in these motor losses. Brown et al[3] found that spine flexibility and upper extremity use were closely related. Spinal fusion (and presumably brace treatment, as well) produced a long lever arm that could not be controlled by the weak hip muscles. The ability to perform activities of daily living independently decreased and more adaptive equipment was required. Despite these recognized risks, Brown et al[3] recommended early surgical stabilization to achieve a stable trunk in adulthood. The majority of our patients reported improved sitting balance and improved endurance after surgery. None regretted having spine stabilization and did not report loss of upper extremity use.

There is a concern about performing major surgery on patients with compromised pulmonary function. The reported complication rates have been variable. Evans et al[8] had no fatalities but did have one case of respiratory obstruction. Daher et al[5] reported a 46% complication rate. More than half of their complications were minor and did not require further surgery. Hensinger and MacEwen[10] had two postoperative deaths, one 5 weeks after surgery and one 10 months later. Three of their 20 surgical patients required grafting for a pseudoarthrosis. Aprin et al[10] studied 22 patients who underwent spine fusion. Atelectasis and pneumonitis developed in 10 of these patients. Four required postoperative intubation for 5 days, and the remainder responded to conservative management. Late complications included weak neck muscles in 4, chest wall narrowing in 6 that developed during postoperative casting, pseudarthrosis in 1, and progression of curves in 5 patients. In our series of nine surgical patients, there were no postoperative deaths related to surgery. There was one pseudarthrosis that required further surgery and one painful hardware that required removal. The patient who underwent subcutaneous rodding has progressed her curve and will require further surgery in the near future. Only one patient had significant respiratory compromise after surgery and required tracheostomy. Interestingly, there were no wound complications in our series or in other reports.

Aggressive physical and respiratory therapy preoperatively and postoperatively can decrease the pulmonary complications.[1,10] Most of the reported series, including our own, used Harrington rods that required postoperative casting and may have contributed to the loss of function as well as chest wall deformity. The use of Luque segmental fixation (Figure 2) and, now, the Cotrel-Dubousset system (Figure 1) has allowed early mobility postoperatively and eliminated the need for casting. Brown et al reported a lower complication rate in patients treated with segmental spinal instrumentation.[3]

In summary, children who survive beyond infancy should be treated with the expectation of prolonged survival. Given this prolonged survival, more aggressive orthopaedic management, including surgical treatment of spinal deformity in the appropriate patient, is justified. Progressive spine deformities that cause progressive medical and functional difficulties develop in children with SMA. It appears that successful arthrodesis prevents more disabling deformity from occurring. Our current management is to surgically stabilize these curves before a severe deformity develops. We agree that bracing can be useful

in the young child to delay the need for surgery, but it does not eliminate the need to surgically stabilize the spine. Correction of deformities can improve function, allowing a child to sit more comfortably, improving handling of the child, and increasing the sitting height.

The low incidence of serious complications also argues that these children can safely be treated surgically when indicated to prevent progression of the deformity and to improve quality of life. Technical improvements in instrumentation may result in a lower incidence of complications since the patients can be mobilized earlier without needing a brace or cast.

In conclusion, we believe that children with SMA should be treated with the expectation of prolonged survival. They should be evaluated regularly to detect the scoliosis that inevitably develops in the most severe forms of SMA. Initial treatment should consist of early bracing. Patients who cannot tolerate brace wear or who have progressive curves require surgical stabilization. The patients and their families need to be well informed about the potential medical complications as well as the possible loss of independent function after surgery.

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